Abstract of the Disclosure

Methods for detecting various types of polymorphic nucleic acid sequences are provided herein. The detection methods are based upon nucleic acid amplification procedures and the ability to detect "large" deletions or insertions in an automated fashion. For example, a deletion or an insertion in a target nucleic acid sequence in a test sample, wherein the deletion or insertion is at least 8 or more consecutive nucleotides, can be detected according to the following steps:

a) contacting the test sample with amplification reagents and a set of amplification primers to form a reaction mixture wherein the set of amplification primers hybridize with the target nucleic acid sequence and a standard nucleic acid sequence in the test sample;

b) subjecting the reaction mixture to amplification conditions to form a target nucleic acid sequence amplification product and a standard nucleic acid amplification product;

c) hybridizing a first labeled probe to the target sequence amplification product and a second labeled probe to the standard nucleic acid sequence amplification product;

d) detecting signals from the first probe and the second probe; and

e) comparing the signals from the first and second labeled probes to determine the presence of the deletion or insertion in the target nucleic acid sequence in the test sample.